Locus 115 (rs12146727)

SNiPAcard

Block annotations

Block info

genomic range	chr12:7,166,770-7,181,948
block size	15,179 bp
variant count	17 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.153 [-3.322 – 3.781]	gene(s) hit or close-by	C1S
phastCons	μ = 0.061 [0 – 1]	eQTL gene(s)	C1RL, C1S
GERP++	μ = -0.675 [-8.63 – 5.15]	potentially regulated gene(s)	C1S, C1S
CADD score	μ = 4.078 [0.316 – 24.8]	disease gene(s)	C1S

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
C1S	COMPLEMENT COMPONENT C1s DEFICIENCY	OMIM	MIM:613783
C1S	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
C1S	missense variant	ENST00000328916	NM_201442.2	ENSP00000328173	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000406697	?	ENSP00000385035	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000403949	?	ENSP00000384464	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000443875	?	ENSP00000397921	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000360817	NM_001734.3	ENSP00000354057	H/R	cAt/cGt	?	?	1
C1S	missense variant	ENST00000617865	?	ENSP00000484657	H/R	cAt/cGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
C1S	ENST00000328916	ILMN_1677603	skin	2.11×10-13 (p-value)	MuTHER consortium	14
C1S	ENST00000488701		adipocyte	1.35×10-7 (p-value)	MuTHER consortium	14
C1S	ENST00000403949					
C1S	ENST00000413211					
C1S	ENST00000443875					
C1S	ENST00000402681					
C1S	ENST00000617865					
C1RL	ENST00000504702	ILMN_1733288	blood	6.73×10-7 (p-value)	Westra et al.	2

C1RL	ENST00000266542

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000129238	1	ENST00000328916, ENST00000360817, ENST00000443875, ENST00000406697, ENST00000402681	C1S

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000015893		C1S
ENCP00000015894		C1S

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001461176 (promoter flanking region)	2	NHLF	H3K36me3
		endothelium (HUVEC)	H3K27me3
		HSMMtube	DNase1
		lung (IMR90)	H3K36me3
		blood (DND-41)	H3K27me3
		skin (NHEK)	H3K4me1
		muscle (HSMM)	H2AZ, H3K27ac, DNase1
ENSR00001609158 (CTCF binding site)	2	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C1S	upstream gene variant, downstream gene variant	602	ENST00000461983	?	?	13
C1S	upstream gene variant, downstream gene variant	226	ENST00000542978	?	ENSP00000442298	7
C1S	upstream gene variant, downstream gene variant	47	ENST00000413211	?	ENSP00000406643	6
C1S	upstream gene variant, downstream gene variant	464	ENST00000360817	NM_001734.3	ENSP00000354057	7
C1S	upstream gene variant, downstream gene variant	38	ENST00000497061	?	?	12
C1S	upstream gene variant, downstream gene variant	144	ENST00000463798	?	?	8
C1S	upstream gene variant, downstream gene variant	447	ENST00000402681	?	ENSP00000384171	7
C1S	upstream gene variant, downstream gene variant	377	ENST00000443875	?	ENSP00000397921	7
C1S	upstream gene variant, downstream gene variant	460	ENST00000473545	?	?	9
C1S	upstream gene variant, downstream gene variant	877	ENST00000495053	?	?	7
C1S	upstream gene variant, downstream gene variant	375	ENST00000328916	NM_201442.2	ENSP00000328173	7
C1S	upstream gene variant, downstream gene variant	54	ENST00000423384	?	ENSP00000399892	6
C1S	upstream gene variant, downstream gene variant	420	ENST00000541647	?	?	5
C1S	upstream gene variant, downstream gene variant	129	ENST00000489541	?	?	7
C1S	upstream gene variant, downstream gene variant	378	ENST00000617865	?	ENSP00000484657	7
C1S	upstream gene variant, downstream gene variant	224	ENST00000543187	?	?	6
C1S	upstream gene variant, downstream gene variant	475	ENST00000495061	?	?	13
C1S	upstream gene variant, downstream gene variant	224	ENST00000488701	?	?	6
C1S	upstream gene variant, downstream gene variant	594	ENST00000470326	?	?	13

C1S	upstream gene variant, downstream gene variant	400	ENST00000403949	?	ENSP00000384464	5
C1S	downstream gene variant	375	ENST00000406697	?	ENSP00000385035	6

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1S	ENST00000463798	?	?	2
C1S	ENST00000402681	?	ENSP00000384171	9
C1S	ENST00000328916	NM_201442.2	ENSP00000328173	8
C1S	ENST00000406697	?	ENSP00000385035	9
C1S	ENST00000360817	NM_001734.3	ENSP00000354057	8
C1S	ENST00000542978	?	ENSP00000442298	5
C1S	ENST00000443875	?	ENSP00000397921	8
C1S	ENST00000461983	?	?	3
C1S	ENST00000423384	?	ENSP00000399892	1
C1S	ENST00000541647	?	?	5
C1S	ENST00000543187	?	?	1
C1S	ENST00000617865	?	ENSP00000484657	8
C1S	ENST00000488701	?	?	1
C1S	ENST00000495053	?	?	4
C1S	ENST00000403949	?	ENSP00000384464	3
C1S	ENST00000473545	?	?	1
C1S	ENST00000413211	?	ENSP00000406643	1
C1S	ENST00000495061	?	?	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1S	ENST00000402681	?	ENSP00000384171	1
C1S	ENST00000328916	NM_201442.2	ENSP00000328173	1
C1S	ENST00000406697	?	ENSP00000385035	1
C1S	ENST00000360817	NM_001734.3	ENSP00000354057	1
C1S	ENST00000443875	?	ENSP00000397921	1
C1S	ENST00000617865	?	ENSP00000484657	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1S	ENST00000402681	?	ENSP00000384171	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
C1S	ENST00000463798	?	1
C1S	ENST00000541647	?	3
C1S	ENST00000495053	?	2

Locus 115 (sentinel SNP rs12146727 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.